



ADVANCING RARE DISEASE RESEARCH:

The Intersection of Patient Registries, Biospecimen Repositories, and Clinical Data

January 11–12, 2010 • Doubletree Hotel & Executive Meeting Center • Bethesda, MD

DAY 1 — MONDAY, JANUARY 11

Please find workshop presentations, speaker abstracts, breakout session background material and summaries, and workshop summaries by clicking on the presentation or breakout session titles below.

Workshop Objectives: *To discuss the development of an infrastructure for an Internet-based platform with common data elements utilizing a federated rare disease registry able to incorporate:*

1. Existing rare disease registries and any other useful patient registries
2. Patient organizations with no registry looking to establish one
3. Patients with no affiliation with a support group looking to belong to a registry

Expected Outcomes: *To gain acceptance of the concept of a federated rare disease patient registry by as many curators of patient registries and other stakeholders as possible and to gain their participation in creating this patient registry. Participating stakeholders will agree on a strategy to harmonize standardized common data elements, vocabulary, and open source software to enable the exchange of data and information to facilitate research collaborations.*

PLENARY SESSIONS

A. Introduction to Objectives for the Collaborative Rare Diseases Registry (CRDR)

Developing the Rare Diseases Registry

Stephen C. Groft, Pharm.D. — ORDR, NIH

Patient and Research Advocate Statement

Amy Farber, Ph.D. — LAM Treatment Alliance

Uniting Rare Diseases

Vanessa Rangel Miller, M.S., C.G.C. — DuchenneConnect



B. Alternatives and Future Promises for a National Rare Disease Patient Registry

Structure and Function of a Collaborative Rare Disease Patient Registry

Christopher B. Forrest, M.D., Ph.D. — University of Pennsylvania School of Medicine and Children's Hospital of Philadelphia

The National Health Information Network and its Implications for a National Rare Disease Patient Registry/Case study

Daniel C. Russler, M.D. — Oracle Health Sciences Strategy

PRESENTATIONS

I. Standards, Informatics, and Technology

Support for Compatibility and Interoperability

Kyle Brown — Innolyst, Inc.

Reuse of Clinical Health Records: Caveat Inquisitor

James J. Cimino, M.D. — Laboratory for Informatics Development, Clinical Center (CC), NIH

Pros and Cons of Various Models and Communication Across the Different Models

Clement J. McDonald, M.D. — National Library of Medicine (NLM), NIH

Plans for Data Standards in Rare Disease Registries

Rachel Richesson, Ph.D., M.P.H. — University of South Florida (USF) College of Medicine

Global Data Aggregation: Case Study/Treat-NMD

Christophe Beroud, Pharm.D., Ph.D. — France, INSERM

II. Biospecimens/Biorepositories

Challenges and Obstacles Obtaining Rare Disease Specimens and the Use of Registries

Christopher A. Moskaluk, M.D., Ph.D. — UVA Biorepository

Rare Disease Biospecimens: Quality and Accessibility Challenges

Carolyn C. Compton, M.D., Ph.D. — Office of Biorepositories and Biospecimen Research (OBBR), National Cancer Institute (NCI)

Rare Disease Biorepositories and Registries: The Need for Collaborative and Novel Approaches

Benjamin M. Greenberg, M.D., M.H.S. — University of Texas Southwestern

The Use of Patient Registries to Increase Procurements of Rare Diseases Biospecimens

Jeffrey A. Thomas — National Disease Research Interchange (NRDI)

Investigator Experience: How Research in Rare Diseases Contributes to Understanding the Pathogenesis of Common Diseases

Marsha A. Moses, Ph.D. — Children's Hospital Boston and Harvard Medical School

Keynote Speaker: Advancing Rare Disease Research: Ethical Dimensions

Jonathan D. Moreno, Ph.D. — David and Lyn Silfen University Professor of Ethics and Professor of Medical Ethics and the History and Sociology of Science at the University of Pennsylvania

III. Clinical Research, Patient Care, and Disease Management

Role of Rare Disease Registries in Clinical Research

Ronald A. Christensen, M.D. — REGISTRAT-MAPI

Regulatory and Other Governmental Influences on Clinical Research

Theresa Toigo, R.Ph., M.B.A. — Office of Special Health Issues, Food and Drug Administration (FDA)

Patient Registries and their Role in Understanding Health Outcomes

Jean R. Slutsky, P.A., M.S.P.H. — Center for Outcomes and Evidence, Agency for Healthcare Research and Quality (AHRQ)

Data and Test Result Validation: Reporting Research Data and Clinical Test Results to Patients (Researcher and Patient Perspectives)

Andrew Faucett, M.S., C.G.C. — Emory University School of Medicine and Collaboration, Education, and Test Translation (CETT) Program

IV. Patient Participation and Outreach Activities/Patient Advocacy

Patient Advocacy Groups and Patient Registries: An Overview

Sukirti N. Bagal, M.D. — National Organization for Rare Disorders (NORD)

The Role of Patient Advocacy Groups in Establishing Common Infrastructure

Sharon F. Terry, M.A. — Genetic Alliance

Essential Elements for Translational Research in Rare Diseases: Progeria as a Case Study

Leslie B. Gordon, M.D., Ph.D. — Progeria Research Foundation

Common Diseases versus Rare Diseases: Is There Really a Difference?

Susan M. Love, M.D. — Dr. Susan Love Research Foundation

Participation of Patients with no Advocacy Group

David S. Goldstein, M.D., Ph.D. — Clinical Neurocardiology Section, National Institute of Neurological Disorders and Stroke (NINDS), NIH

V. Human Subjects: Bioethical and Legal Issues for Clinical Studies

Human Subjects: Ethical and Legal Issues/45 CFR 46

Julie Kaneshiro, M.A. — Office for Human Research Protections (OHRP)

Ethical and Legal Issues/Government Regulations

P. Pearl O'Rourke, M.D. — Partners Healthcare

Legal/Bioethical Issues in Medical Research and Release of Genetic Information

Jack Schwartz, J.D. — University of Maryland School of Law



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DAY 2 — TUESDAY, JANUARY 12

Keynote Speaker: The Contribution of Large Health Care Systems to Improving Treatment for Patients with Rare Diseases

Joe V. Selby, M.D., M.P.H. — Division of Research, Kaiser Permanente

BREAKOUT SESSIONS

Morning Parallel Sessions Introduction and Instructions

Moderator: *Helen M. Moore, Ph.D.* — Office of Biospecimen and Biorepositories Research, NCI

A. Standardized Vocabulary, Terminology, CDE's, and Diagnosis

Chairs: *Rachel Richesson, Ph.D., M.P.H.* and *Clement J. McDonald, M.D.*

Note taker: *Kyle Brown*

Discussion Panel:

Clement J. McDonald, M.D. — NLM, NIH

Rachel Richesson, Ph.D., M.P.H. — USF College of Medicine

Stuart Nelson, M.D., F.A.C.M.I. — NLM, NIH

Michael S. Watson, M.S., Ph.D. — American College of Medical Genetics

George A. Komatsoulis, Ph.D. — Center for Biomedical Informatics and Information Technology (CBIIT), NCI

Kyle Brown — Innolyst, Inc.

B.1 Patient Participation/Outreach Activities and Patient Advocacy

Chairs: *Amy Farber, Ph.D., Kate McCurdy, and Paul A. Harris, Ph.D.*

Note takers: *Kate McCurdy and Jennifer Farmer, M.S., C.G.C.*

Discussion Panel:

Ronald J. Bartek — FARA

Jennifer Farmer, M.S., C.G.C. — FARA

Amy Farber, Ph.D. — LAM Treatment Alliance

Leslie B. Gordon, M.D., Ph.D. — Progeria Research Foundation

Lynn Etheredge — Rapid Learning Project, George Washington University

Paul A. Harris, Ph.D. — Office of Research Informatics Operation, Vanderbilt University



C.1 Biospecimens/Biorepositories

Chairs: *Jim B. Vaught*, Ph.D. and *Christopher A. Moskaluk*, M.D., Ph.D.

Note takers: *Josh Sommer*

Discussion Panel:

Jim B. Vaught, Ph.D. — OBBR, NCI

Christopher A. Moskaluk, M.D., Ph.D. — UVA Biorepository

Simone S. Sommer, M.D., M.P.H. — Chordoma Foundation

Sharon F. Terry, M.A. — Genetic Alliance

Benjamin M. Greenberg, M.D., M.H.S. — University of Texas Southwestern

Ian M. Fore, D.Phil. — CBIIT, NCI

Jeffrey A. Thomas — NDRI

Afternoon Parallel Sessions

B2. Clinical Trials/Research Studies and Patient Care Management

Chairs: *Ronald A. Christensen*, M.D. and *Vanessa Rangel Miller*, M.S., C.G.C.

Note takers: *Rachel Richesson*, Ph.D., M.P.H. and *Kate McCurdy*

Discussion Panel:

Ronald A. Christensen, M.D. — REGISTRAT-MAPI

Vanessa Rangel Miller, M.S., C.G.C. — DuchenneConnect

Christopher B. Forrest, M.D., Ph.D. — University of Pennsylvania School of Medicine and Children's Hospital of Philadelphia

Robert H. Shelton, M.B.A. — Private Access, Inc.

Santa J. Tumminia, Ph.D. — National Eye Institute (NEI), NIH

Dianne M. Finkelstein, Ph.D. — Harvard University

C2. Human Subjects: Bioethical and Legal Issues

Chairs: *Jack Schwartz*, J.D. and *Sara C. Hull*, Ph.D.

Note taker: *Amy Farber*, Ph.D. and *Jennifer Farmer*, M.S., C.G.C. — FARA

Discussion Panel:

Jack Schwartz, J.D. — University of Maryland School of Law

Julie Kaneshiro, M.A. — OHRP

Sara C. Hull, Ph.D. — National Human Genome Research Institute (NHGRI), NIH

P. Pearl O'Rourke, M.D. — Partners Healthcare

Barbara I. Karp, M.D. — NINDS, NIH

Wendy E. Patterson — Technology Transfer Center, NCI

D. Informatics/Database Technology

Chairs: *Kyle Brown* and *Lisa Forman-Neall*, Ph.D.

Note taker: *Rachel Richesson*, Ph.D., M.P.H.

Discussion Panel:

Kyle Brown — Innolyst, Inc.

Lisa Forman-Neall, Ph.D. — National Center for Biotechnology Information (NCBI), NLM

Christophe Beroud, Pharm.D., Ph.D. — France, INSERM

Rachel Richesson, Ph.D., M.P.H. — USF College of Medicine

Chalapathy Neti, Ph.D., B.S. — IBM Research

Workshop Summary

Day 1: *Ronald A. Christensen*, M.D. and *Christopher B. Forrest*, M.D., Ph.D.

Day 2: Breakout session presentations by the chairs of each session — summary, recommendations, and action items

ADDITIONAL MATERIAL

[ORDR/NHGRI Genetic and Rare Diseases \(GARD\) Information Center](#)

[FDA Orphan Drugs Development](#)